

UNIVERSITY OF WASHINGTON

SEATTLE, WASHINGTON 98105

February 17, 1971

School of Medicine
Department of Medicine
DIVISION OF MEDICAL GENETICS

Dr. Victor A McKusick
Division of Medical Genetics
The Johns Hopkins Hospital
Baltimore, Md. 21205

Dear Dr. McKusick:

Thank you for the program of this summer's Birth Defects Conference.

As I mentioned to you recently, Dr. Motulsky and I, in collaboration with Ed Bierman of the Division of Metabolism, have begun several studies on the genetics of the hyperlipoproteinemias and their relationship to coronary artery disease. Other than for the information available on familial hypercholesterolemia (Type II), there is no solid genetic data available on these disorders. Hopefully, our planned studies will provide a better understanding of the familial hypertriglyceridemias as well as an unbiased documentation of how much coronary artery disease is associated with the genetic hyperlipoproteinemias. The results of these studies, unfortunately, will not be ready for presentation this summer.

However, one of our side projects is to study a large Type II kindred in the Aleutian Islands. Our field trip to Alaska is planned for March 17, 1971, and if all goes as expected I'll have some interesting and new data to report. The kindred we plan to study lives as an isolated group in the Aleutian Islands and arose from the mating of an Aleut (affected) and a German male (unaffected). In contrast to the large kindred with Type II reported by Harlan, Estes, and Graham in Medicine in 1964, the affected members of the Alaska family are said to have severe and premature coronary artery disease. Thus, formal genetic and linkage studies will obviously be of interest.

If agreeable with you, I thought I would present at the Baltimore Conference data from our Aleutian expedition, emphasizing the clinical characteristics in those affected family members under age 20 and comparing this family with the Harlan family and with Kachadurian's Lebanese isolate. If the USPHS Hospital in Alaska has any autopsy data on members of the Aleutian kindred, I will plan to include this in my presentation. What do you think?

I look forward to seeing you in Atlantic City in May. I am very anxious to discuss with you some of my recent observations on Alstrom's Syndrome (3 sisters) and on a family with dominantly inherited sexual ateliotic dwarfism in whom growth hormone, glucose, insulin, and lipid responses are all normal !

Sincerely yours,

Joseph L. Goldstein, M. D.